CLAIMS

What is claimed is:

1. A method for diagnosis of a disorder associated with the development of beta amyloid deposits or fibrils in a human or animal subject or assessing the efficacy of treatment rendered to the subject for such disorder, said method comprising the step of:

A) determining the presence of mtDNA CR mutations.

2. A method according to Claim 1, wherein Step A comprises making a qualitative determination that mtDNS CR mutation is or is not present.

3. A method according to Claim 1, wherein Step A comprises making a quantitative determination of mtDNS CR mutations.

4. A method according to Claim 3 further comprising the step of:

B) comparing a mtDNS CR value obtained by the quantitative determination made in Step A with a control mtDNS CR value to determine whether the subject has significantly more mtDNS CR mutations than control.

5. A method according to Claim 3 further comprising the step of:

B) comparing a mtDNS CR value obtained by the quantitative determination made in Step A with a mtDNS CR value representative of subjects who suffer from a disorder associated with the development of beta amyloid deposits or fibrils.

6. A method according to any of Claim 1 wherein Step A comprises testing for a T4141G mutation.

7. A method according to any of Claim 1 wherein Step A comprises testing for a T414C mutation.

8. A method according to any of Claim 1 wherein Step A comprises testing for a T477C mutation.

9. A method according to any of Claim 1 wherein Step A comprises testing for a T146C mutation.

10. A method according to any of Claim 1 wherein Step A comprises testing for a T152C mutation.

11. A method according to any of Claim 1 wherein Step A comprises testing for a A189G mutation.

12. A method according to any of Claim 1 wherein Step A comprises testing for a T195C mutation.

13. A method according to Claim 1 wherein Step A is carried out at least in part by PNA-clamping PCR.

14. A method according to Claim 1 wherein Step A is carried out at least in part by oligonucleotide hybridization.

15. A method according to Claim 1 wherein Step A is carried out at least in part by primer extension.

16. A method according to Claim 1 wherein Step A is carried out at least in part by restriction digestion.

17. A method according to Claim 1 wherein the determination of Step A is made in a specimen of tissue, cells or body fluid selected from the group consisting of:

i. brain tissue;
ii. brain tissue from the frontal cortex;
iii. nervous tissue;
iv. nerve cells

v. blood
vi. blood cells;
vii. urine;
viii. urinary tract cells;
ix. skin;
x. skin cells;
xi. epithelium;
xii. epithelial cells;
xiii. fibroblasts;
xiv. cerebrospinal fluid; and
xv. cells contained in cerebrospinal fluid.

18. A method according to Claim 1 wherein the method is carried out for post-symptomatic diagnosis of a disorder in a subject who has begun to exhibit symptoms of that disorder.

19. A method according to Claim 1 wherein the method is carried out for pre-symptomatic diagnosis of a disorder in a subject who has not begun to exhibit symptoms of that disorder.

20. A method according to Claim 1 wherein the disorder is a neurodegenerative disease.

21. A method according to Claim 1 wherein the disorder is Alzheimer's Disease.

22. A method according to Claim 1 wherein the disorder is Parkinson's Disease.

23. A method according to Claim 1 wherein the disorder is Down's Syndrome-associated dementia.

24. A method according to Claim 1 wherein the disorder is a spongiform encephalopathy.

25. A method according to Claim 1 wherein the disorder is type II diabetes.

26. A method according to Claim 1 wherein the disorder is Creutzfeldt-Jakob disease.

27. A method according to Claim 1 wherein the disorder is a Huntington's disease.

28. A method according to Claim 1 wherein the disorder is macular degeneration.

29. A method according to Claim 1 wherein the disorder is a prion disease.

30. A method according to Claim 1 wherein Step A comprises:

obtaining sample cells from the subject;

extracting DNA from the sample cells;

subjecting the extracted DNA to mitochondrial DNA control region amplification;

determining whether homoplasmic 414 and 477 nucleotide variants are present by direct sequencing for heteroplasmic 414 and 477 nucleotide mutations; and

if 414 and 477 nucleotide variants are detected, cloning the mutant molecules and sequencing the clone.

31. A test system comprising reagents and/or materials useable to perform a method according to any of Claims 1-30.

32. A test system according to claim 31 further comprising instructions for use.

33. A test system according to claim 31 further comprising a reference containing control data.

34. A test system according to claim 33 wherein the reference comprises computer software.